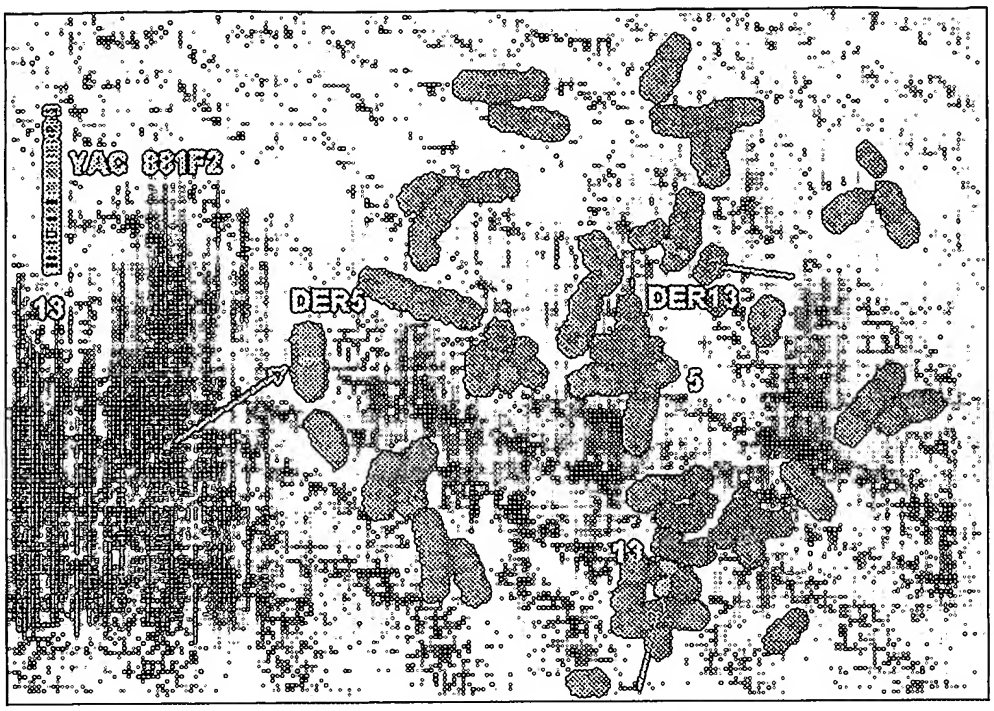
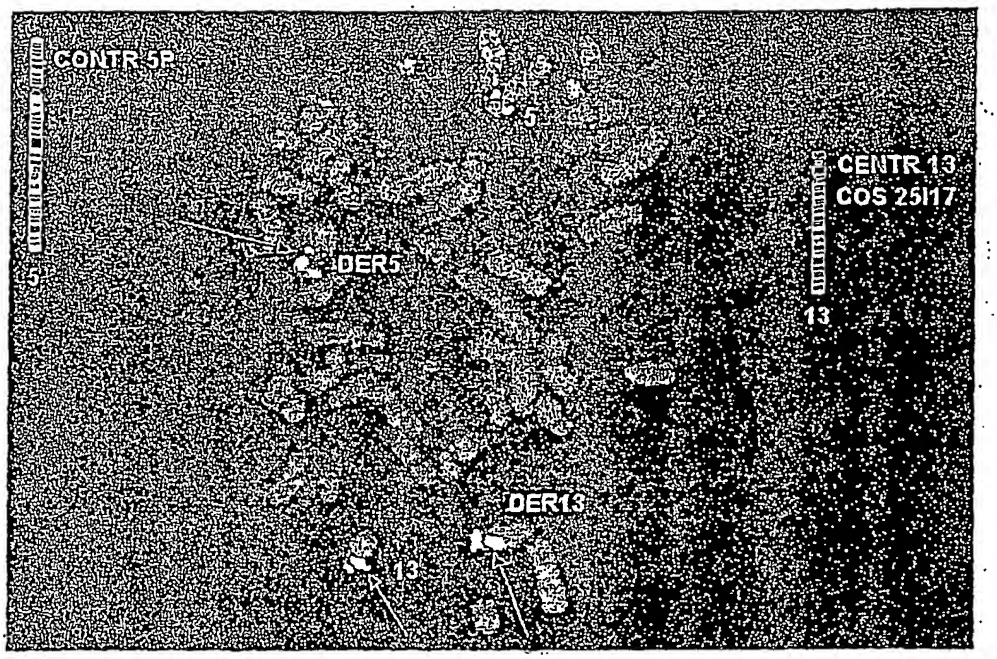
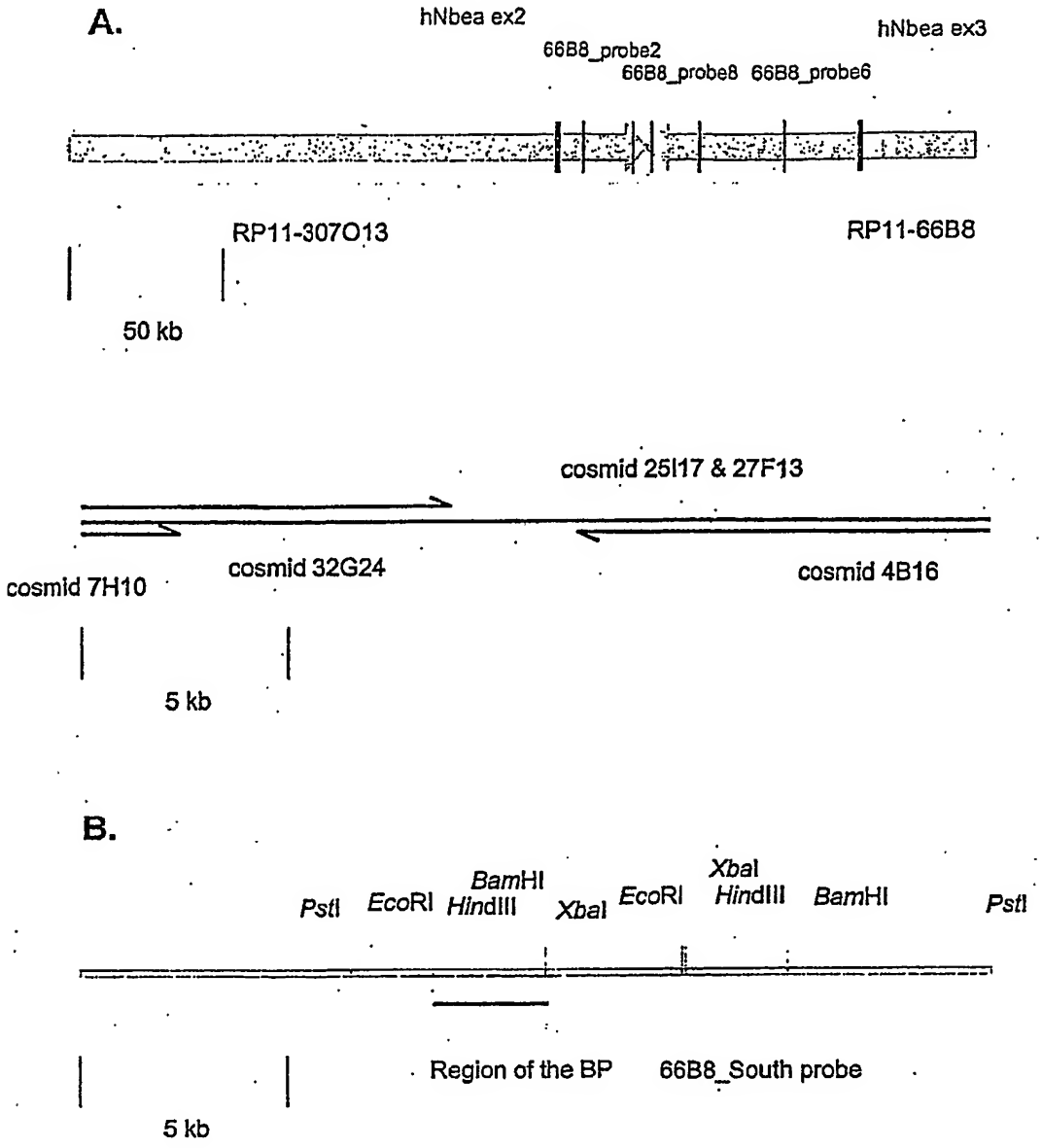


Figure 1. FISH analysis of metaphase chromosomes of VC cells of the autistic patient with probe YAC 881f2. Hybridization signals are marked by arrows with a chromosome identity is label.



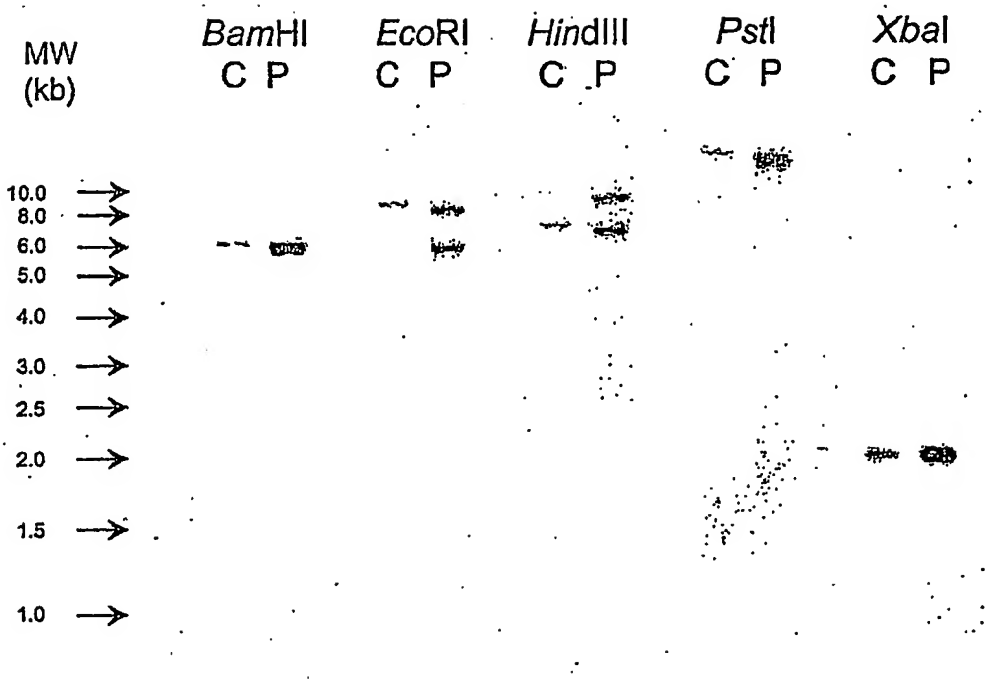
~~Figure 2.~~ FISH analysis of metaphase chromosomes of VC cells of the autistic patient using cosmid clone 25I17. Hybridization signals are marked by arrows with a chromosome identity is label.

Figure 1.



~~Figure 3 :~~ Map of the breakpoint (BP) region in the autistic patient. (A) FISH analysis. At the BAC level, clone 307O13 is proximal and 66B8 distal to the BP. At the level of cosmids (obtained by cosmid library screening with 66B8_probe2, 6 and 8), cosmids 32G24 and 7H10 are proximal, and 4B16 is distal to the BP. Cosmids 25I17 and 27F13 span the BP. (B) Restriction map used for Southern blot analysis with 66B8_South probe. This analysis (see figure) narrowed down the BP region to a 2.8 kb *Hin*dIII/*Bam*HI restriction fragment.

Figure 2.



~~Figure 4.~~ Southern blot analysis of genomic DNA from the autistic patient (P) and a control individual (C). Genomic DNA was digested with the mentioned restriction enzymes. Using probe 66B8_South, rearranged fragments are observed for the *Eco*RI (5.9 kb), *Hin*dIII (8.7 kb) and *Pst*I (11.5 kb) digestions in the patient, in addition to the 8.0 kb, 6.4 kb and 14.7 kb wild type fragments , respectively. In the control, only the wild type fragment is visible.

Figure 3

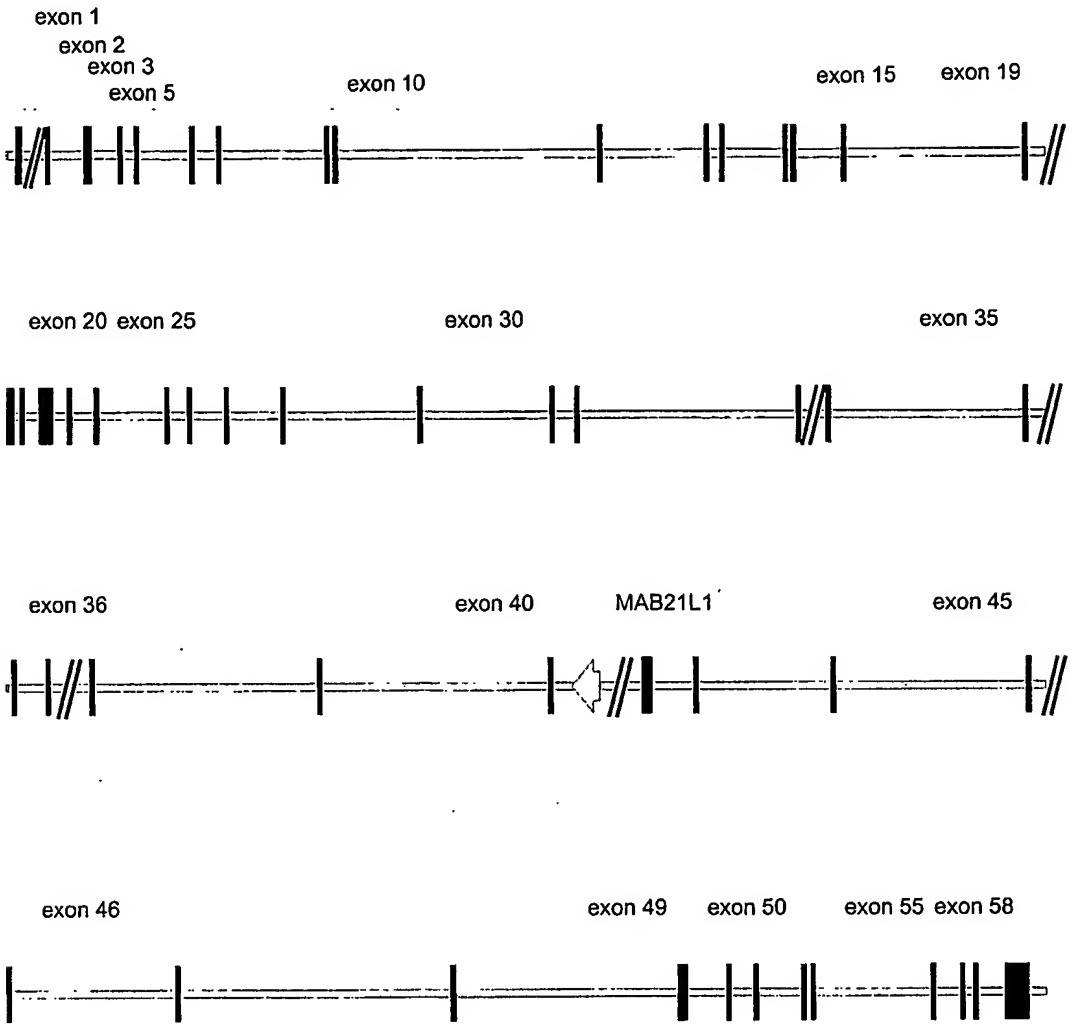


Figure 5 . Genomic structure of hNbea. Exons and introns of hNbea are depicted to scale, except some huge introns (>55 kb) that are truncated (//). The unique exon of the MAB21L gene is also represented.

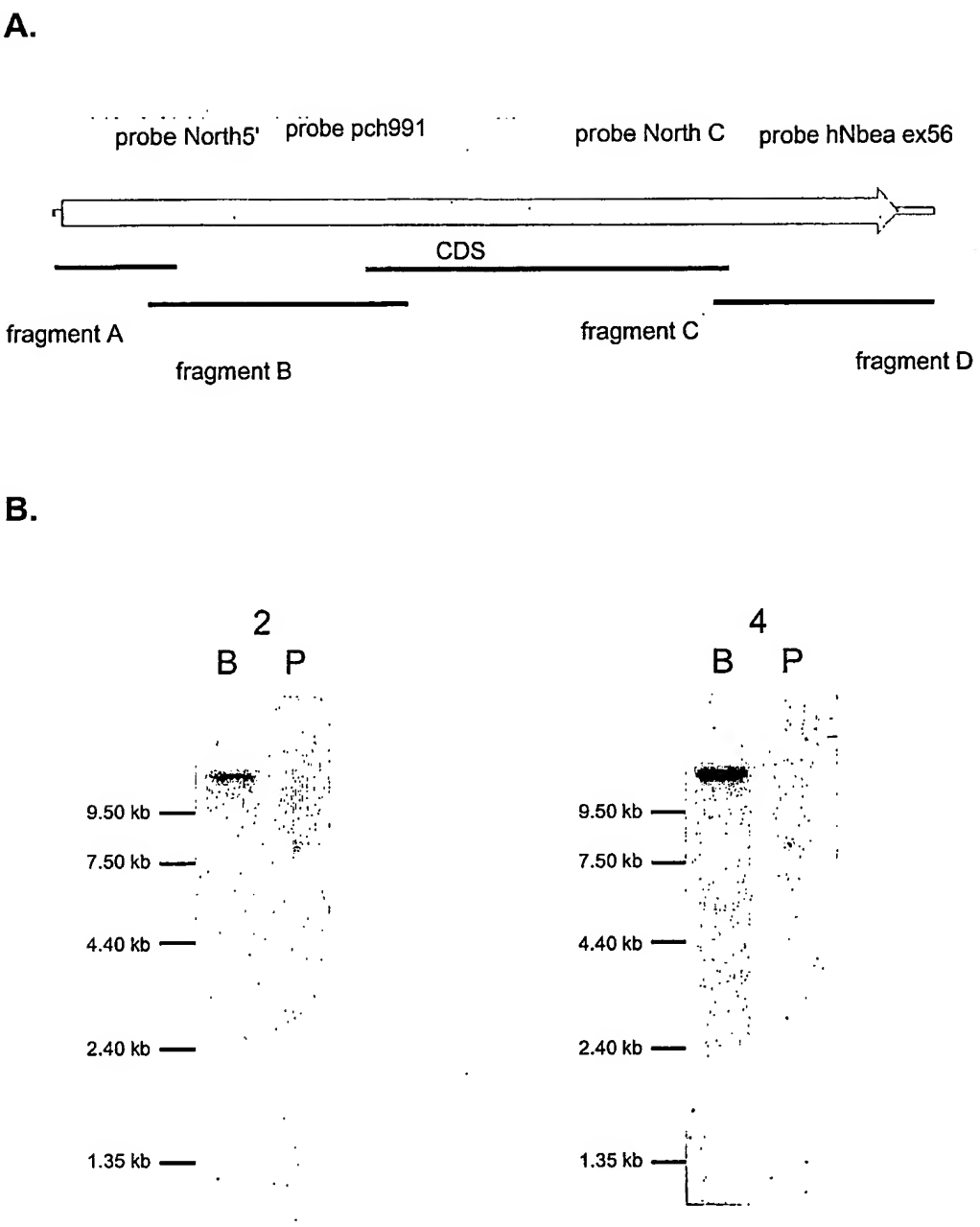


Figure 6. Northern blot analysis of the hNbea transcript. (A) Schematic representation of the assembled cDNA sequence showing the four fragments (A to D) cloned separately by RT-PCR and the probes used for Northern analysis. (B) Northern blots of total RNA from human tissues (Clontech). Probes used for hybridization are: 1, North5'; 2, pch991; 3, NorthC; 4, hNbea-ex56. Tissue abbreviations are: B, brain; P, placenta.

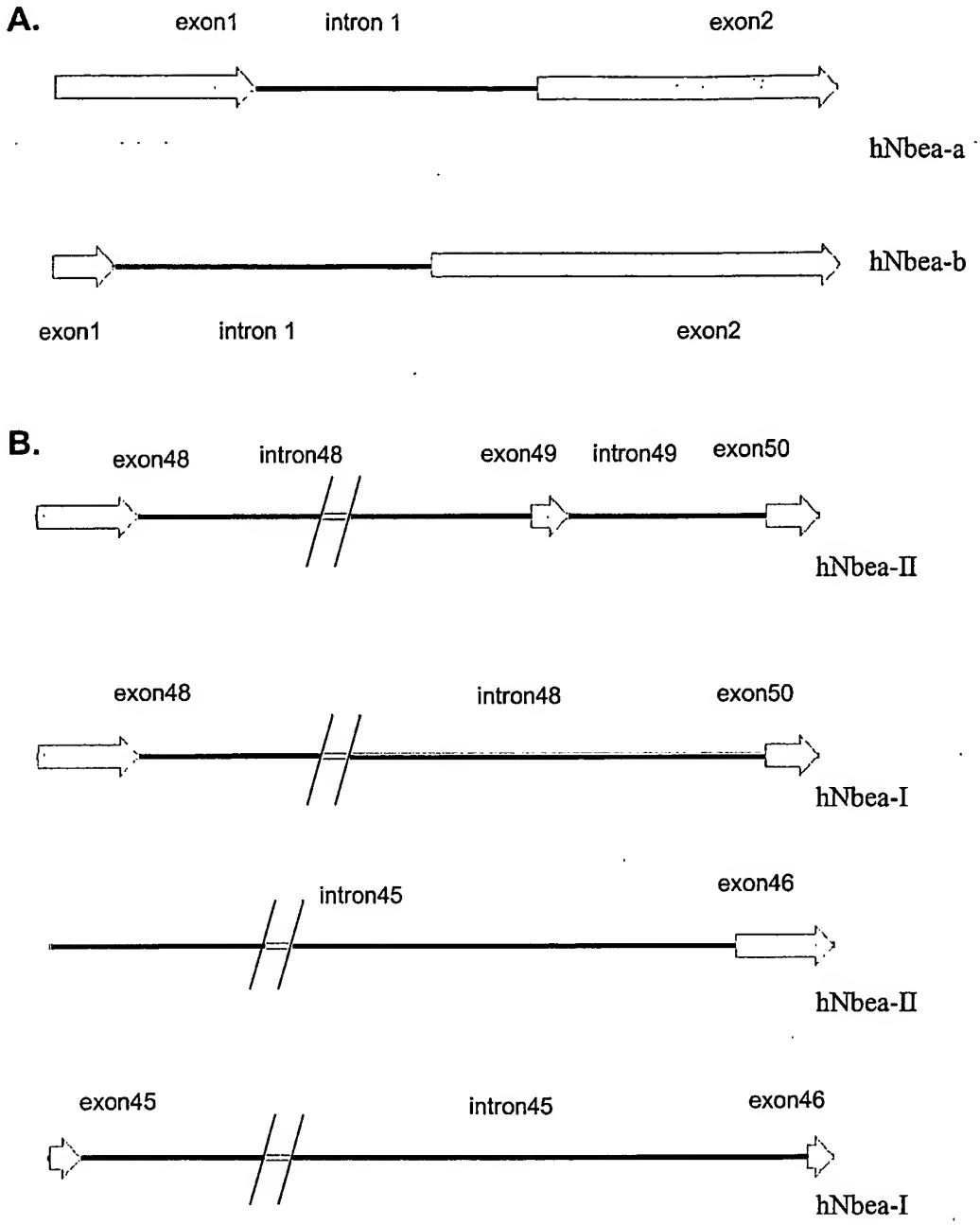
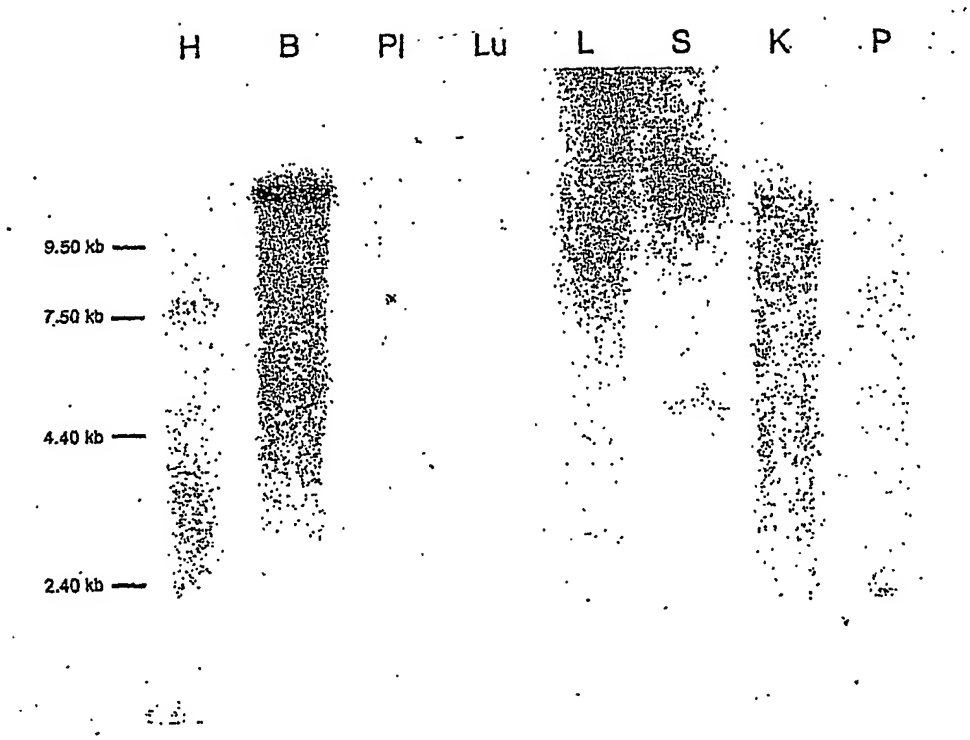


Figure 7. Splicing variants of hNbea. (A) hNbea-I; two variants (hNbea-a and -b) of exon 1 and 2 leading to differences in the 15 first aa . (B) Differences between hNbea-I and -II; the additional exon 49, and the extended initial exon 46 in hNbea-II.



~~Figure 8:~~ **Expression pattern of the hNbea transcript.** Northern blots of total RNA from human tissues (*Clontech*). The probe used for hybridization is hNbea-ex56. Tissue abbreviations are as followed: B, brain; H, heart; K, kidney; L, liver; Lu, lung; P, pancreas; Pl, placenta; S, skeletal muscle.

Figure 4

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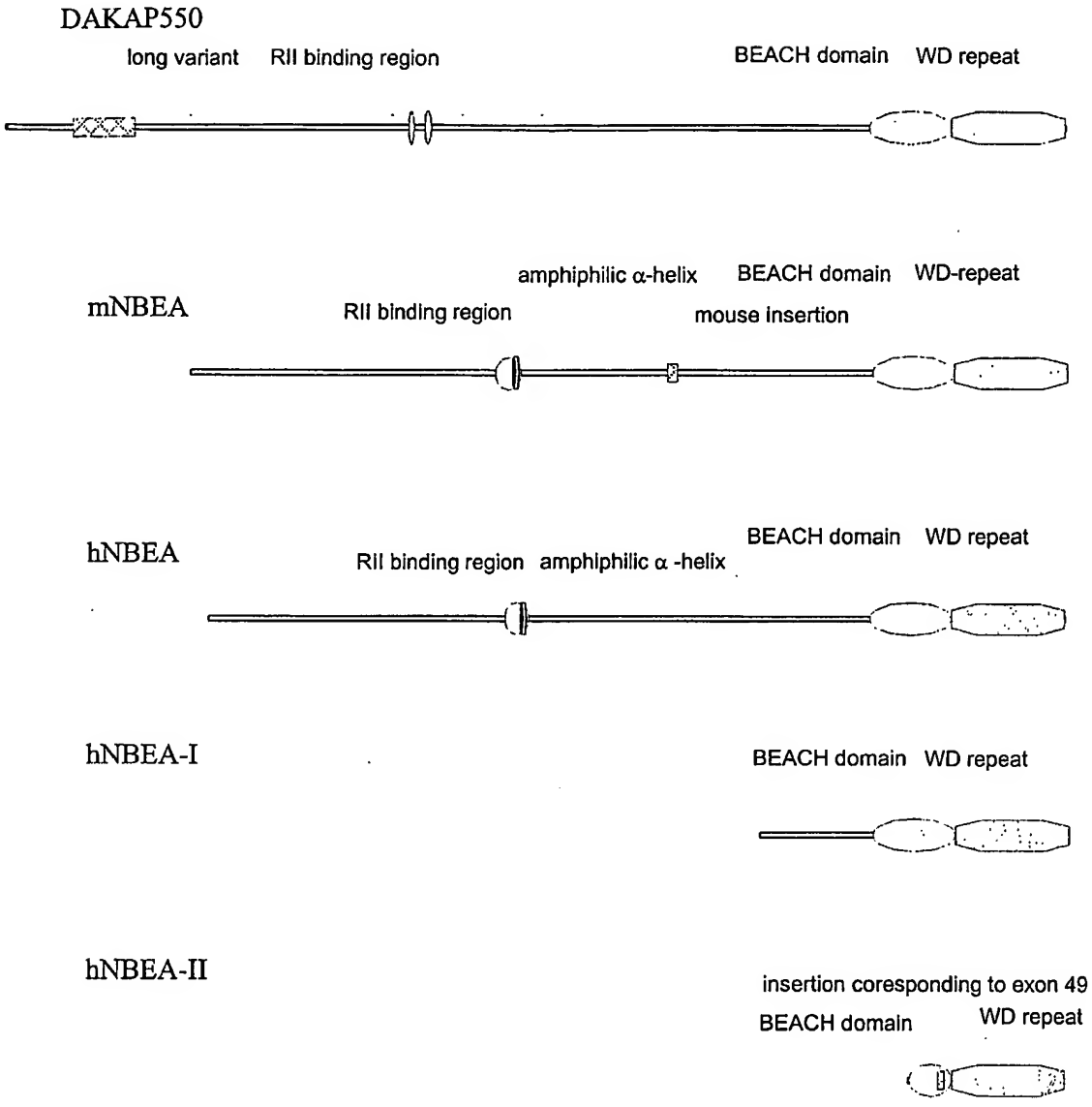

Figure 10 . Graphical representation of neurobeachin proteins. BEACH-WD40 module was recognized *in silico*. The site for RII-PKA binding was demonstrated experimentally for mNBEA (Wang *et al.*, 2000) and DAKAP550 (Han *et al.*, 1997). In the human homologue the corresponding region is 100% identical to that of mNBEA. Putative amphiphilic α–helix are depicted within this PKA binding domain.